

# Recombinant Human GAA therapeutic protein(Alglucosidase alfa)

**Cat. No.** GAA-P026H    **Lot. No.** (See product label)

## SPECIFICATION

**Product Overview**

Recombinant Human acid alpha-glucosidase therapeutic protein consists of the human enzyme acid alpha-glucosidase (GAA) which is essential for the degradation of glycogen to glucose in lysosomes. It is encoded by the most predominant of nine observed haplotypes of this gene. It is produced by recombinant DNA technology in a Chinese hamster ovary cell line. The protein degrades glycogen by catalyzing the hydrolysis of  $\alpha$ -1,4- and  $\alpha$ -1,6- glycosidic linkages of lysosomal glycogen. Structurally, it is a glycoprotein with a calculated mass of 98,008 daltons for the 883 residue mature polypeptide chain, and a total mass of approximately 109,000 daltons, including carbohydrates. It is used for the treatment of Pompe disease (GAA deficiency) in infants and pediatric patients.

**Species**                      Human

**Source**                        CHO

**Description**

This gene encodes lysosomal alpha-glucosidase, which is essential for the degradation of glycogen to glucose in lysosomes. The encoded preproprotein is proteolytically processed to generate multiple intermediate forms and the mature form of the enzyme. Defects in this gene are the cause of glycogen storage disease II, also known as Pompe's disease, which is an autosomal recessive disorder with a broad clinical spectrum. Alternative splicing results in multiple transcript variants.

**Molecular Mass**            105270.802

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**AA Sequence**

AHPGRPRAVPTQCDVPPNSRFDKAITQEQCEARGCCYIPAKQGLQGAQMGG  
 PWCFFPPSYPSYKLEN LSSSEMGYTATLTRTPTFFPKDILTRLDMMETENRLHF  
 TIKDPANRRYEVPLETPHVHSRAPSPLYSV EFSEEPFGVIVRRQLDGRVLLNTTVAP  
 LFFADQFLQLSTSLPSQYITGLAEHLSPLMLSTSWTRITLWNRD LAPTPGANLYGSH  
 PFYLALEDGGS AHGVFLNNSAMDVVLQPSPALSWRSTGGILDVYIFLGPEPKSVVQ  
 Q YLDVVGYPFMPYWG LGFHLCRWGYSSAITRQVVENMTRAHFPLDVQWNDLD  
 YMDSRRDFTFNKDGFRDF PAMVQELHQGGRRYMMIVDPAISSSGPAGSYRPHYDE  
 GLRRGVFITNETGQPLIGKVWPGSTAFP DFT NPTALAWWEDMVAEFHDQVPFDGM  
 WIDMNEPSNFIRGSEDGCPNNELENPPYVPGVVG GTLQAATICASSH QFLSTHYNL  
 HNLYGLTEAIASHRALVKARGTRPFVISRSTFAGHG RYAGHWTGDVWSSWEQLASS  
 VPEILQ FNLLGVPLVGADVCGFLGNTSEELCVRWTQLGAFYPFMRNHNSLLSLPQE  
 PYSFSEPAQQAMRKALTRYA LLPHYTLFHQAHVAGETVARPLFLEFPKDSSTWT  
 VDHQLLWGEALLITPVLQAGKAEVTGYFPLGTWYDL QTVPVEALGSLPPPPAAPRE  
 PAIHSEGQWVTL PAPLDTINVHLRAGYIIP LQGPGLTTTESRQQPMALAVA LTKGGE  
 ARGELFWDDGESLEVLERGAYTQVIFLARNTIVNELVRVTSEGAGLQLQKVTVLGV  
 ATAP QQVLSNGVPVSNFTYSPDTKVL DICVSLLMGEQFLVSWC

**Endotoxin** < 0.1 EU per µg of the protein

**Purity** >95%

**Alias** GAA; LYAG; Alglucosidase alfa

## GENE INFORMATION

**Gene Name** GAA glucosidase, alpha; acid [ Homo sapiens ]

**Official Symbol** GAA

**Synonyms** GAA; glucosidase, alpha; acid; lysosomal alpha-glucosidase; glycogen storage

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	disease type II; Pompe disease; acid maltase; aglucosidase alfa; LYAG;
<b>Gene ID</b>	<a href="#">2548</a>
<b>mRNA Refseq</b>	<a href="#">NM_000152</a>
<b>Protein Refseq</b>	<a href="#">NP_000143</a>
<b>MIM</b>	<a href="#">606800</a>
<b>UniProt ID</b>	<a href="#">P10253</a>
<b>Chromosome Location</b>	17q25.2-q25.3
<b>Pathway</b>	Galactose metabolism, organism-specific biosystem; Galactose metabolism, conserved biosystem; Lysosome, organism-specific biosystem; Lysosome, conserved biosystem; Metabolic pathways, organism-specific biosystem; Notch-mediated HES/HEY network, organism-specific biosystem; Starch and sucrose metabolism, organism-specific biosystem;
<b>Function</b>	alpha-glucosidase activity; carbohydrate binding; hydrolase activity, hydrolyzing O-glycosyl compounds; maltose alpha-glucosidase activity;

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